



PKP2 gene

plakophilin 2

Normal Function

The *PKP2* gene provides instructions for making a protein called plakophilin 2. This protein is found primarily in cells of the myocardium, which is the muscular wall of the heart. Within these cells, plakophilin 2 is one of several proteins that make up structures called desmosomes. These structures form junctions that attach cells to one another. Desmosomes provide strength to the myocardium and are involved in signaling between neighboring cells.

Health Conditions Related to Genetic Changes

arrhythmogenic right ventricular cardiomyopathy

More than 50 mutations in the *PKP2* gene have been identified in people with arrhythmogenic right ventricular cardiomyopathy (ARVC). This condition most commonly affects the myocardium surrounding the right ventricle, one of the two lower chambers of the heart. ARVC increases the risk of an abnormal heartbeat (arrhythmia) and sudden death.

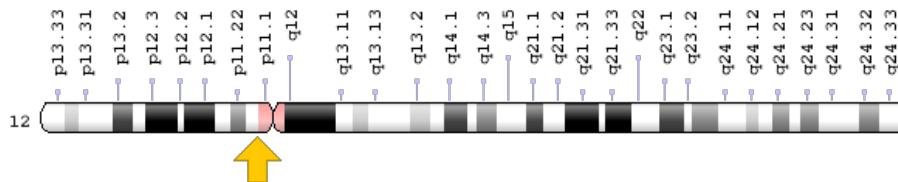
Some *PKP2* gene mutations lead to the production of an abnormally short version of plakophilin 2. Other mutations alter the structure of plakophilin 2 by adding, deleting, or changing one or more of its protein building blocks (amino acids). Studies suggest that the altered protein impairs the formation and function of desmosomes.

Without normal desmosomes, cells of the myocardium detach from one another and die, particularly when the heart muscle is placed under stress (such as during vigorous exercise). The damaged myocardium is gradually replaced by fat and scar tissue. As this abnormal tissue builds up, the walls of the right ventricle become stretched out, preventing the heart from pumping blood effectively. These changes also disrupt the electrical signals that control the heartbeat, which can lead to arrhythmia.

Chromosomal Location

Cytogenetic Location: 12p11.21, which is the short (p) arm of chromosome 12 at position 11.21

Molecular Location: base pairs 32,790,746 to 32,896,846 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARVD9
- MGC177501
- plakophilin-2

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Desmosomes Connect Intermediate Filaments from Cell to Cell
<https://www.ncbi.nlm.nih.gov/books/NBK26857/#A3488>
- Molecular Cell Biology (first edition, 2000): Desmosomes
<https://www.ncbi.nlm.nih.gov/books/NBK21599/#A6512>

GeneReviews

- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
<https://www.ncbi.nlm.nih.gov/books/NBK1131>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PKP2%5BTIAB%5D%29+OR+%28plakophilin+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- PLAKOPHILIN 2
<http://omim.org/entry/602861>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PKP2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PKP2%5Bgene%5D>
- HGNC Gene Family: Armadillo repeat containing
<http://www.genenames.org/cgi-bin/genefamilies/set/409>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9024
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5318>
- UniProt
<http://www.uniprot.org/uniprot/Q99959>

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